X-linked severe combined immunodeficiency

X-linked severe combined immunodeficiency (SCID) is an inherited disorder of the immune system that occurs almost exclusively in males. Boys with X-linked SCID are prone to recurrent and persistent infections because they lack the necessary immune cells to fight off certain bacteria, viruses, and fungi. Many infants with X-linked SCID develop chronic diarrhea, a fungal infection called thrush, and skin rashes. Affected individuals also grow more slowly than other children. Without treatment, males with X-linked SCID usually do not live beyond infancy.

Frequency

X-linked SCID is the most common form of severe combined immunodeficiency. Its exact incidence is unknown, but the condition probably affects at least 1 in 50,000 to 100,000 newborns.

Genetic Changes

Mutations in the *IL2RG* gene cause X-linked SCID. The *IL2RG* gene provides instructions for making a protein that is critical for normal immune system function. This protein is necessary for the growth and maturation of developing immune system cells called lymphocytes. Lymphocytes defend the body against potentially harmful invaders, make antibodies, and help regulate the entire immune system. Mutations in the *IL2RG* gene prevent these cells from developing and functioning normally. Without functional lymphocytes, the body is unable to fight off infections.

Inheritance Pattern

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- IL2RG SCID, T- B+ NK-
- SCIDX1

- X-linked SCID
- X-SCID
- XSCID

Diagnosis & Management

Formal Diagnostic Criteria

- ACT Sheet: Severe Combined Immunodeficiency (SCID) and Conditions Associated with T Cell Lymphoneia
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/SCID.pdf

Genetic Testing

- Genetic Testing Registry: X-linked severe combined immunodeficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1279481/

Other Diagnosis and Management Resources

- Baby's First Test: Severe Combined Immunodeficiency
  http://www.babysfirsttest.org/newborn-screening/conditions/severe-combined-immunodeficiency-scid
- GeneReview: X-Linked Severe Combined Immunodeficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1410
- MedlinePlus Encyclopedia: Immunodeficiency Disorders
  https://medlineplus.gov/ency/article/000818.htm
- National Marrow Donor Program: Severe Combined Immunodeficiency and Transplant
  https://bethematch.org/for-patients-and-families/learning-about-your-disease/severe-combined-immunodeficiency/

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Immunodeficiency Disorders
  https://medlineplus.gov/ency/article/000818.htm
- Health Topic: Immune System and Disorders
  https://medlineplus.gov/immunesystemanddisorders.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- X-linked severe combined immunodeficiency
  https://rarediseases.info.nih.gov/diseases/5618/x-linked-severe-combined-immunodeficiency

Additional NIH Resources

- National Human Genome Research Institute: Learning About Severe Combined Immunodeficiency
  https://www.genome.gov/13014325/
- National Institute of Allergy and Infectious Diseases: Primary Immune Deficiency Diseases
  https://www.niaid.nih.gov/diseases-conditions/primary-immune-deficiency-diseases-pidds

Educational Resources

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/severe-combined-immunodeficiency-scid
- Genetic Science Learning Center, University of Utah
  http://learn.genetics.utah.edu/content/disorders/singlegene/
- Great Ormond Street Hospital for Children NHS Trust (UK)
  http://www.gosh.nhs.uk/medical-information-0/search-medical-conditions/severe-combined-immunodeficiency-scid
- KidsHealth from the Nemours Foundation
  http://kidshealth.org/en/parents/severe-immunodeficiency.html

- Merck Manual Consumer Version
  http://www.merckmanuals.com/home/immune-disorders/immunodeficiency-disorders/severe-combined-immunodeficiency-scid
- Orphanet: T-B+ severe combined immunodeficiency due to gamma chain deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=276

Patient Support and Advocacy Resources

- Immune Deficiency Foundation
  http://primaryimmune.org/
- International Patient Organisation for Primary Immunodeficiencies
  http://www.ipopi.org/
- Jeffrey Modell Foundation
  http://www.info4pi.org/
- National Organization for Rare Disorders
  https://rarediseases.org/rare-diseases/severe-combined-immunodeficiency/

GeneReviews

- X-Linked Severe Combined Immunodeficiency
  https://www.ncbi.nlm.nih.gov/books/NBK1410

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22x-linked+severe+combined+immunodeficiency%22+OR+%22severe+combined+immunodeficiency%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28Severe+Combined+Immunodeficiency%5BMAJR%5D%29+AND+%28%28x-linked+severe+combined+immunodeficiency%5BTIAB%5D%29+OR+%28scidx1%5BTIAB%5D%29+OR+%28x-scid%5BTIAB%5D%29+OR+%28xscid%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SEVERE COMBINED IMMUNODEFICIENCY, X-LINKED
  http://omim.org/entry/300400

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